# General

## Guideline Title

Genetic considerations for a woman's pre-conception evaluation.

## Bibliographic Source(s)

Wilson RD, Audibert F, Brock JA, Cartier L, Desilets VA, Gagnon A, Johnson JA, Langlois S, Murphy-Kaulbeck L, Okun N, Pastuck M. Genetic considerations for a woman's pre-conception evaluation. J Obstet Gynaecol Can. 2011 Jan;33(1):57-64. [31 references] PubMed

## Guideline Status

This is the current release of the guideline.

# Recommendations

## Major Recommendations

Recommendations and Summary Statements

A review of the current literature does not provide enough information for this committee opinion to present evidence-based recommendations.

Summary

Pre-conception planning is important but may not always be possible because some pregnancies are unintended. The reproductive risks must be considered at all patient interactions because personal situations change, and new discoveries can affect family histories. Primary prevention of genetic and congenital anomalies is the goal in reducing perinatal morbidity and mortality and enhancing healthy families. Health care providers should always ask patients of reproductive age "Are you considering a pregnancy or could you possibly become pregnant?"

Table: Taking a Pre-Conception History for Assessment and Counselling

Genetic History

A thorough pre-conception history identifies couples who are genetically at risk. When women and their partners are informed of the risks of having a baby with birth defects or a genetic disorder prior to pregnancy, they are then able to determine their options regarding a pregnancy (including contraception, artificial insemination, adoption, prenatal invasive testing, or chance).

*Family History*

- Construct three-generation pedigree.

Table: Taking a Pre-Conception History for Assessment and Counselling

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| • Include assessment of genetic diseases, including muscular dystrophy, hemophilia, cystic fibrosis, fragile X syndrome, congenital heart disease, phenylketonuria, dwarfism, sickle cell anemia, and Tay-Sachs disease.<br>• Include assessment of multifactorial congenital malformations, such as spina bifida, anencephaly, cleft palate and cleft lip, hypospadias, and congenital heart disease.<br>• Include assessment of familial diseases with a major genetic component, such as developmental disability, premature atherosclerosis, diabetes mellitus, psychosis, epileptic disorders, hypertension, rheumatoid arthritis, deafness, and severe refractive disorders of the eye.<br><br>*Ethnic History*<br><br>• Establish risk for specific conditions related to ethnic origin, such as sickle cell anemia, Tay-Sachs disease, neural tube defects, beta-thalassemia, and alpha-thalassemia.<br><br>*Age*<br><br>• Establish risks associated with age (e.g., women under age 15 or over age 35 may carry increased biological risks). |
| Health History<br><br>*Chronic Conditions*<br><br>• Assess the presence of chronic conditions that can affect a woman's ability to conceive, as well as the use of medications in treatment of chronic disease and their potential effect on pregnancy.<br>• The following specific conditions should be considered: diabetes mellitus, anemias, thyroid disorders, gynaecological disorders, hyperphenylalaninemia, asthma, sexually transmitted infections, heart disease, hypertension, deep venous thrombosis, kidney disease, systemic lupus erythematosus, epilepsy, hemoglobinopathies, cancer, seizure disorders, tuberculosis, rheumatoid arthritis, and mental health/psychiatric disorders.<br><br>*Infectious Conditions*<br><br>• Identify women who are rubella or varicella susceptible. If they are not actively attempting pregnancy, offer a vaccination.<br>• Identify and counsel women at risk for hepatitis B. Routine pre-conception testing of all women with hepatitis B is not currently recommended.<br>• Counsel women to avoid exposure to cat feces and raw and undercooked meats. Routine serologic testing for toxoplasmosis in the pre-conception period is not recommended.<br>• Evaluate the woman and her partner for exposure to sexually transmitted disease (e.g., Chlamydia, human immunodeficiency virus [HIV], gonorrhea, syphilis).<br><br>*Reproductive History*<br><br>• Collect information about menstrual, contraceptive, and sexual histories; infertility; abnormal Pap smears; and in utero exposure to diethylstilbestrol.<br>• Discuss past obstetric history, including early miscarriages; number of pregnancies; type of birth; length of labour; and specific complications, such as premature labour or delivery, gestational diabetes, pregnancy-induced hypertension, and postpartum depression.<br>• Discuss menstrual difficulties, specifically excessive cyclic bleeding, amenorrhea, and oligomenorrhea.<br>• Discuss gynaecological disease, such as endometriosis and pelvic inflammatory disease.<br><br>*Lifestyle Assessment*<br><br>• Assess lifestyle issues, including nutrition; physical activity; prescription and over-the-counter drug use; other substance use; and environmental exposures, current and past. |

Adapted from Public Health Agency of Canada. Family-centred maternity and newborn care: national guidelines. Chapter 3. Preconception care. Health Canada, 2005.

## Clinical Algorithm(s)

None provided

# Scope

## Disease/Condition(s)

- Birth defects
- Genetic and congenital anomalies

## Guideline Category

Counseling

Prevention

Risk Assessment

Screening

## Clinical Specialty

Family Practice

Internal Medicine

Medical Genetics

Obstetrics and Gynecology

Preventive Medicine

## Intended Users

Advanced Practice Nurses

Allied Health Personnel

Health Care Providers

Physician Assistants

Physicians

## Guideline Objective(s)

To give health care providers information about the genetic information that can be used as part of health surveillance for women undergoing a pre-conception evaluation for genetic risk assessment and possible genetic screening or testing

## Target Population

Women of reproductive age and their partners

## Interventions and Practices Considered

Pre-conception evaluation for genetic risk assessment and possible genetic screening or testing

### Major Outcomes Considered

- Maternal, infant, and child health outcomes
- Risk of genetic and congenital anomalies

## Methodology

### Methods Used to Collect/Select the Evidence

Searches of Electronic Databases

Searches of Unpublished Data

### Description of Methods Used to Collect/Select the Evidence

PubMed or Medline and the Cochrane Database were searched in November 2009, using appropriate key words (preconception, genetic disease, maternal, family history, genetic health risk, genetic health surveillance, prenatal screening, prenatal diagnosis, birth defects, and teratogen). Grey (unpublished) literature was identified through searching the websites of health technology assessment and health technology assessment-related agencies, clinical practice guideline collections, and national and international medical specialty societies.

### Number of Source Documents

Not stated

### Methods Used to Assess the Quality and Strength of the Evidence

Expert Consensus (Committee)

### Rating Scheme for the Strength of the Evidence

Not applicable

### Methods Used to Analyze the Evidence

Review

### Description of the Methods Used to Analyze the Evidence

Not stated

### Methods Used to Formulate the Recommendations

Expert Consensus

### Description of Methods Used to Formulate the Recommendations

Not stated

### Rating Scheme for the Strength of the Recommendations

Not applicable

### Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

### Method of Guideline Validation

Internal Peer Review

### Description of Method of Guideline Validation

This committee opinion has been prepared and reviewed by the Genetics Committee, reviewed by the MD Advisory Committee and the Medico-Legal Committee and approved by the Executive of the Society of Obstetricians and Gynaecologists of Canada.

## Evidence Supporting the Recommendations

### Type of Evidence Supporting the Recommendations

The type of evidence supporting the recommendations is not specifically stated.

## Benefits/Harms of Implementing the Guideline Recommendations

### Potential Benefits

The benefits for the patient and her family include understanding of possible genetic risk and enhanced pregnancy outcomes.

### Potential Harms

The harms include increased anxiety or psychological stress associated with the possibility of identifying genetic risks.

## Qualifying Statements

### Qualifying Statements

This document reflects emerging clinical and scientific advances on the date issued and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed. Local institutions can dictate amendments to these opinions. They should be well documented if modified at the local level. 

## Implementation of the Guideline

### Description of Implementation Strategy

An implementation strategy was not provided.

# Institute of Medicine (IOM) National Healthcare Quality Report Categories

## IOM Care Need

Staying Healthy

## IOM Domain

Effectiveness

Patient-centeredness

# Identifying Information and Availability

## Bibliographic Source(s)

Wilson RD, Audibert F, Brock JA, Cartier L, Desilets VA, Gagnon A, Johnson JA, Langlois S, Murphy-Kaulbeck L, Okun N, Pastuck M. Genetic considerations for a woman's pre-conception evaluation. J Obstet Gynaecol Can. 2011 Jan;33(1):57-64. [31 references] PubMed

## Adaptation

Not applicable: The guideline was not adapted from another source.

## Date Released

2011 Jan

## Guideline Developer(s)

Society of Obstetricians and Gynaecologists of Canada - Medical Specialty Society

## Source(s) of Funding

Society of Obstetricians and Gynaecologists of Canada

## Guideline Committee

Genetics Committee

## Composition of Group That Authored the Guideline

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## Financial Disclosures/Conflicts of Interest

Disclosure statements have been received from all members of the committee.

## Guideline Status

This is the current release of the guideline.

## Guideline Availability

Electronic copies: Available in Portable Document Format (PDF) from the Society of Obstetricians and Gynaecologists (SOGC) of Canada Web site .

Print copies: Available from the Society of Obstetricians and Gynaecologists of Canada, La société des obstétriciens et gynécologues du Canada (SOGC) 780 promenade Echo Drive Ottawa, ON K1S 5R7 (Canada); Phone: 1-800-561-2416.

## Availability of Companion Documents

None available

## Patient Resources

None available

## NGC Status

This NGC summary was completed by ECRI Institute on April 6, 2011. The information was verified by the guideline developer on May 10, 2011.

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